ALDH5A1 gene

aldehyde dehydrogenase 5 family member A1

Normal Function

The *ALDH5A1* gene provides instructions for producing the succinic semialdehyde dehydrogenase enzyme. This enzyme is found in the energy-producing centers of cells (mitochondria). Succinic semialdehyde dehydrogenase is involved in the breakdown of a chemical that transmits signals in the brain (neurotransmitter) called gamma-amino butyric acid (GABA). The primary role of GABA is to prevent the brain from being overloaded with too many signals. Once GABA molecules have been released from nerve cells, they are broken down by succinic semialdehyde dehydrogenase and other enzymes.

Health Conditions Related to Genetic Changes

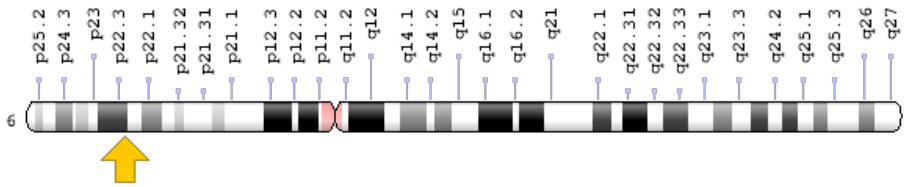
succinic semialdehyde dehydrogenase deficiency

At least 35 mutations in the *ALDH5A1* gene have been found to cause succinic semialdehyde dehydrogenase deficiency. Most of these mutations change one protein building block (amino acid) in the succinic semialdehyde dehydrogenase enzyme. Mutations in the *ALDH5A1* gene lead to the production of an enzyme with little or no activity. A lack of functional succinic semialdehyde dehydrogenase disrupts the conversion of succinic semialdehyde to succinic acid. Instead, succinic semialdehyde is converted back into GABA or to a related molecule, gamma-hydroxybutyrate (GHB). It is unclear how increases in GHB and GABA cause developmental delay, seizures, and other features of succinic semialdehyde dehydrogenase deficiency.

Chromosomal Location

Cytogenetic Location: 6p22.3, which is the short (p) arm of chromosome 6 at position 22.3

Molecular Location: base pairs 24,494,969 to 24,537,207 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- aldehyde dehydrogenase 5 family, member A1
- aldehyde dehydrogenase 5 family, member A1 (succinate-semialdehyde dehydrogenase)
- aldehyde dehydrogenase 5A1
- mitochondrial succinate semialdehyde dehydrogenase
- NAD(+)-dependent succinic semialdehyde dehydrogenase
- SSADH
- SSDH
- SSDH_HUMAN

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): GABA shunt reactions are responsible for the synthesis, conservation and metabolism of GABA https://www.ncbi.nlm.nih.gov/books/NBK27979/?rendertype=figure&id=A1180
- Basic Neurochemistry (sixth edition, 1999): GABA Synthesis, Uptake and Release https://www.ncbi.nlm.nih.gov/books/NBK27979/

GeneReviews

- Succinic Semialdehyde Dehydrogenase Deficiency https://www.ncbi.nlm.nih.gov/books/NBK1195

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALDH5A1%5BTIAB%5D%29+OR+%28%28SSDH%5BTIAB%5D%29+OR+%28SSADH%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

OMIM

- ALDEHYDE DEHYDROGENASE 5 FAMILY, MEMBER A1
  http://omim.org/entry/610045

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH5A1%5Bgene%5D
- HGNC Gene Family: Aldehyde dehydrogenases
  http://www.genenames.org/cgi-bin/genefamilies/set/398
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=408
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7915
- UniProt
  http://www.uniprot.org/uniprot/P51649

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